

NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
May 21-22, 2018

The Open Session of the 83rd meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, May 21, 2018, at the Fishers Lane Terrace Level Conference Center in Rockville, Maryland. Dr. Eric Green, Director of the National Human Genome Research Institute (NHGRI), called the meeting to order.

The meeting was open to the public from 10:00 AM until 5:00 PM on May 21, 2018. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 8:00 AM to 10:00 AM and 5:00 PM to 5:30 PM on May 21, 2018, and from 8:30 AM until adjournment on May 22, 2018, for the review, discussion, and evaluation of grant applications.

Council Members Present:

Carol Bult
Brenton Graveley
Gail Henderson
Mark Johnston
Jonathan Haines
Jonathan Pritchard*
Dan Roden*
Val Sheffield
Jay Shendure

Ad Hoc Advisors Present

Jeffrey Botkin
Wendy Chung
Patricia Deverka
Stephen Fodor
Trey Ideker
Rafael Irizarry
Sharon Plon*
Aviv Regev*
*Indicates participation by phone

Staff Present from the National Institutes of Health:

Gabriel Batzli, ERP	Ann Fitzpatrick, DM
Vivien Bonazzi, OD	Valentina di Francesco, ERP
Vence Bonham, IOD and IRP	Sheena Faherty, DPCE
Joy Boyer, ERP	Elise Feingold, ERP
Comfort Browne, ERP	Adam Felsenfeld, ERP
Christine Chang, ERP	Robert Fullem, ERP
Monika Christman, ERP	Tina Gatlin, ERP
Michelle Cohen, ERP	Bettie Graham, ERP
Heather Colley, ERP	Sylvia Garvey, ERP
Erin Currey, ERP	Margaret Ginoza, ERP
Christina Daulton, DPCE	Sarah Gould, ERP
Jyoti Dayal, ERP	Linda Hall, ERP
Carla Easter, DPCE	Lucia Hindorff, ERP

Rebecca Hong, DPCE
Ellen Howerton, ERP
Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Sonya Jooma, DPCE
Sheethal Jose, ERP
Melpi Kasapi, ERP
David Kaufman, ERP
Jimmie Kees, ERP
Britny Kish, DPCE
Jennifer Krupp, NIH-ACMG
Rongling Li, ERP
Nicole Lockhart, ERP
Jon Lorsch, NIGMS
Allison Mandich, IOD
Ebony Madden, ERP
Keith McKenney, ERP
Donna Messersmith, DPCE
Jenn Montooth, DPCE
Sam Moore, ERP
Ken Nakamura, ERP
Vivian Ota Wang, ERP

Michael Pagan, ERP
Kiara Palmer, DPCE
Mike Pazin, ERP
Lita Proctor, ERP
Erin Ramos, ERP
Robb Rowley, ERP
Laura Rodriguez, DPCE
Ben Ryan, IOD
Laura Skow, ERP
Mike Smith, ERP
Heidi Sofia, ERP
Taylorlyn Stephan, ERP
Jeff Struewing, ERP
Barbara Thomas, ERP
Elizabeth Tuck, DPCE
Lu Wang, ERP
Cara Weismann, DPCE
Chris Wellington, ERP
Kris Wetterstrand, IOD
Ken Wiley, ERP
Anthony Williams, ERP
Rosann Wise, DPCE

Others Present for All or a Portion of the Meeting:

Rhonda Schonberg, National Society of Genetic Counselors

Materials, including the agenda and PowerPoint presentations for the Open Session of the 83rd NACHGR meeting are available at: <https://www.genome.gov/27571447/may-2018-council-agenda-and-documents/>

A recorded video of the entire Open Session is available at:

<https://www.youtube.com/playlist?list=PL1ay9ko4A8slwRuBCWmoLNtmUrFn7WdsK>

INTRODUCTION OF NEW NHGRI COUNCIL MEMBERS, STAFF, LIAISONS, AND GUESTS

Dr. Rudy Pozzatti began the Open Session with the introduction of new Council Members, NHGRI staff, liaisons, and guests, which can be found at:

<https://www.youtube.com/watch?v=KKZXmrOy68A&t=0s&list=PL1ay9ko4A8slwRuBCWmoLNtmUrFn7WdsK&index=2>

APPROVAL OF MINUTES FOR THE FEBRUARY 2018 NACHGR MEETING

The Council approved the Minutes by unanimous vote once amended to include Jonathan Haines as an attendee at the February, 2018 Council meeting.

FUTURE MEETING DATES

- Sept. 24-25, 2018
- Feb. 11-12, 2019
- May 20-21, 2019
- Sept. 16-17, 2019

DIRECTOR'S REPORT

Dr. Eric Green gave his Director's Report, which included a series of updates about recent activities at NHGRI, NIH, and in the broader genomics community. A video of his presentation can be found at: <https://www.youtube.com/watch?v=BpnVDeKdGY8>

PRESENTATION – *NIH's Strategic Plan for Data Science*

Dr. Jon Lorsch introduced the NIH Strategic Plan for Data Science developed by the NIH Scientific Data Council. The plan focuses on modernizing the data resource ecosystem to increase its utility for researchers and other stakeholders by modernizing infrastructure, increasing capacity, and enhancing data sharing, access, and interoperability. Dr. Lorsch described the plan's five overarching goals and their strategic objectives, the implementation tactics that will be employed, and the milestones and performance measures that will be used. A video of this presentation can be found at: <https://www.youtube.com/watch?v=OW6gpbcmjNo>

CONCEPT CLEARANCE – *Center for ELSI Resources & Analysis*

Dr. Nicole Lockhart presented a concept titled, *Center for ELSI Resources and Analysis*. This Center will serve to: provide ELSI researchers with an established platform to share their research tools related to genomics, serve as a resource for curating and synthesizing research, and convene ELSI researchers to develop a more synergistic and integrated ELSI research community. The Council voted unanimously to approve the concept. A video of this presentation can be found at: <https://www.youtube.com/watch?v=2iotUe8Ah5o>

CONCEPT CLEARANCE – *Genomic Innovator Award*

Dr. Lisa Brooks presented a concept titled, *Genomic Innovator Award*. This concept will support highly innovative genomic research performed by creative investigators who participate in consortia or similar large research groups and are early in their career development path. Following Council's recommendation, the language will be modified to be more inclusive to those who participate in team science. Council voted unanimously to approve the concept. A video of this presentation can be found at: <https://www.youtube.com/watch?v=BSS3BLk5sME>

PRESENTATION – *Report on the NHGRI Intramural Research Program*

Dr. Dan Kastner, Director of NHGRI's Division of Intramural Research reported on the status of the NHGRI Intramural Research Program (IRP). Dr. Kastner provided the background and history of the IRP, which serves in part to catalyze the "genomic transformation" of the intramural research programs of other NIH Institutes, and to act as an incubator for programs that could be exported to the broader genomics community. Dr. Kastner highlighted several IRP investigators and their research projects. A video of this presentation can be found at: <https://www.youtube.com/watch?v=MhdxFWT0vvM>

REPORT – Update on the Genome Sequencing Program

Dr. Adam Felsenfeld and Mr. Chris Wellington gave an update on the Genome Sequencing Program (GSP). Dr. Felsenfeld explained the structure of the GSP, which is comprised of four components: Centers for Mendelian Genomics (CMGs), Centers for Common Disease Genomics (CCDGs), GSP Analysis Centers, and GSP Coordinating Center. Mr. Wellington highlighted the successes of Phase I of the CMGs and provided information about future work, including continued collaboration with the Knock Out Mouse Project (KOMP). Dr. Felsenfeld discussed the ongoing analyses at the CCDGs and summarized the goals and plans developed from the recent GSP meeting, as well as advice from the GSP External Advisory panel. A video of this presentation can be found at: <https://www.youtube.com/watch?v=q278SPV2WwM>

REPORT – Update on the Undiagnosed Diseases Network

Dr. Anastasia Wise gave an update about the Undiagnosed Diseases Network (UDN). Dr. Wise summarized the objectives of the UDN, which are: improve the level of diagnosis and care for patients with undiagnosed diseases, facilitate research on the etiology of undiagnosed diseases, and promote an integrated and collaborative community to investigate these diseases. Dr. Wise provided an overview of the current UDN and explained plans for the second phase of the UDN, for which awards are anticipated to be made in the fall of 2018. A video of this presentation can be found at: <https://www.youtube.com/watch?v=0xzIZYswy-A>

COUNCIL-INITIATED DISCUSSION

A video of the Council-initiated discussion can be found at:
<https://www.youtube.com/watch?v=7sRKKCToTks>

ANNOUNCEMENTS AND ITEMS OF INTEREST

Two reports from NHGRI's national liaison societies were provided:
[National Society of Genetic Counselors Quarterly Update](#) 
[American Society of Human Genetics Report to Council](#) 

CONFIDENTIALITY AND CONFLICT OF INTEREST

Dr. Pozzatti read the Confidentiality and Conflict of Interest Policy to Council and asked the members to sign the forms provided to them.

REVIEW OF APPLICATIONSⁱ

In the Closed Session, the NACHGR reviewed 205 applications, requesting \$106,022,104 (total cost). The applications included: 97 research project applications (R01, R03, R15, R21 or RM1); 2 research center applications (U54); 59 cooperative agreement applications (U01 or U24); 8 career development or career transition applications (K99/R00, K01 or K08); 8 research education applications (R25); 5 conference applications (R13); 1 institution training application (T32); 15 SBIR Phase I applications (R43); 8 SBIR Phase II applications (R44); and 2 STTR Phase 1 applications (R41); A total of 152 applications totaling \$86,165,571 were recommended by the NACHGR.

This Council Meeting Summary document was prepared by Sarah Gould, Scientific Program Analyst at NHGRI.

09/27/2018

Date

Rudy Pozzatti

Rudy Pozzatti, Ph.D.

Executive Secretary

National Advisory Council for Human Genome Research

09/27/2018

Date

Eric D. Green

Eric Green, M.D, Ph.D.

Chairman

National Advisory Council for Human Genome Research

This report was approved by the NACHGR on September 24, 2018.

ⁱFor the record, it is noted that to avoid a conflict of interest, Council members absent themselves from the meeting when the Council discusses applications from their respective institutions or in which a conflict of interest may occur. Members are asked to sign a statement to this effect. This does not apply to "en bloc" votes.